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New, Earlier Tests Seen Better at Fetal Down Syndrome Detection

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New tests in the first trimester of pregnancy are better at identifying fetuses with Down syndrome than standard tests done later in pregnancy, according to a government-financed study.

The first-trimester tests detected as many as 87 percent of fetuses with the extra chromosome that causes Down syndrome, compared with 81 percent found by tests in the second trimester, the authors write in today's issue of *The New England Journal of Medicine*.

Down syndrome affects more than two million people worldwide, causing physical and mental disabilities. It occurs about once in 700 to 900 live births.

The study, conducted at 15 hospitals and involving more than 38,000 women, is the government's most comprehensive effort to help expectant couples best determine their risk of having a baby with Down syndrome, the world's leading cause of developmental disability.

The study's findings suggest that parents will have a better opportunity to seek an abortion earlier in gestation, Dr. Joe Leigh Simpson, a Baylor College obstetrics and gynecology professor, wrote in an editorial accompanying the study.

"Pregnancy terminations are earlier, more private and far safer than in the second trimester," Dr. Simpson said. The maternal death rate for first-trimester abortions is 1.1 per 100,000 abortions, compared with 7 to 10 per 100,000 in the second trimester, Dr. Simpson wrote.

Advance knowledge of Down syndrome and other birth defects could also help couples prepare, even if they choose not to abort, government doctors said.

The typical screening for Down syndrome is a blood test, conducted 15 to 18 weeks into the pregnancy. The newer alternative, conducted at 11 to 13 weeks' gestation, is a combination blood and ultrasound test that measures the size of the fluid gap in the fetus neck.

The new tests are in use already. The study being published today culminates a \$15 million government-financed effort to determine which tests work best.

Some doctors question research that makes it easier for parents to decide to abort fetuses with Down syndrome.

"I am personally saddened that so many parents believe that a diagnosis of Down syndrome is a reason to terminate a pregnancy," said Dr. Len Leshin, a pediatrician in Corpus Christi, Tex., who has two sons, one with Down syndrome.

The new tests are big advances from the 1970's, when only 25 percent to 30 percent of Down syndrome cases were detected during pregnancy, Dr. Simpson wrote.

But the tests have their limits, said the study's main author, Dr. Fergal Malone, chairman of obstetrics and gynecology at the Royal College of Surgeons in Ireland.

The measurement of the fluid gap in the fetus's neck "is actually a very, very tricky thing to do right," since it requires a technician to measure a space in fractions of a millimeter on an ultrasound computer image, Dr. Malone said. In about 7.5 percent of cases in the study, technicians could not accurately measure the space.